Marfan Syndrome Diagnostic Criteria Checklist


MIM 154700

NAME _______________________________

DATE _______________________________

EXAMINER _______________________________

Index case:
Major criteria in 2 different organ systems
AND involvement of a third organ system.

Relative of index case:
1 major criterion in family history
AND 1 major criterion in an organ system
AND involvement in second organ system.

SKELETAL

Major (Presence of at least 4 of the following manifestations)

__ pectus carinatum
__ pectus excavatum requiring surgery
__ reduced upper to lower segment ratio (Note 1)
__ OR arm span to height ratio >1.05
__ Height _____ Arm span _____ Upper segment _____ Lower segment _____
__ wrist (Note 2) and thumb (Note 3) signs
__ scoliosis of >20° or spondylolisthesis
__ reduced extension at the elbows (<170°)
__ medial displacement of the medial malleolus causing pes planus
__ protrusio acetabulae of any degree (ascertained on radiographs)

Minor

__ pectus excavatum of moderate severity
__ joint hypermobility
__ high arched palate with crowding of teeth
__ facial appearance
__ dolichocephaly,
__ malar hypoplasia,
__ enophthalmos,
__ retrognathia,
__ down-slanting palpebral fissures

__ INVOLVEMENT: 2 major criteria or 1 major and 2 minor
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OCULAR

Major
__ ectopia lentis

Minor
__ flat cornea
__ increased axial length of the globe
__ hypoplastic iris OR hypoplastic ciliary muscle causing decreased miosis

__ INVOLVEMENT: 2 minor criteria

CARDIOVASCULAR

Major
__ dilatation of the ascending aorta with or without aortic regurgitation and involving at least the sinuses of Valsalva
__ dissection of the ascending aorta

Minor
__ mitral valve prolapse with or without mitral valve regurgitation
__ dilatation of the main pulmonary artery, in the absence of valvular or peripheral pulmonic stenosis below the age of 40 years
__ calcification of the mitral annulus below the age of 40 years
__ dilatation or dissection of the descending thoracic or abdominal aorta below age of 50 years

__ INVOLVEMENT: 1 minor criterion

PULMONARY

Minor (only)
__ spontaneous pneumothorax
__ apical blebs

__ INVOLVEMENT: 1 minor criterion

SKIN AND INTEGUMENT

Minor (only)
__ striae atrophicae
__ recurrent or incisional hernia

__ INVOLVEMENT: 1 minor criterion
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DURA

Major
___ lumbosacral dural ectasia by CT or MRI

FAMILY/GENETIC HISTORY

Major
___ first degree relative who independently meets the diagnostic criterion.
NAME _______________________
___ presence of mutation in FBN1 known to cause Marfan syndrome
___ presence of haplotype around FBN1 inherited by descent and unequivocally associated with diagnosed Marfan syndrome in the family

For Differential Diagnosis:
- Homocystinuria - obtain plasma amino acids
- Congenital Contractural Arachnodactyly (MIM 121050)
- Familial Thoracic Aortic Aneurysm (MIM 132900)
- Familial Aortic Dissection (MIM 132900)
- Familial Ectopia Lentis (MIM 129600)
- Familial Marfan-Like Habitus (MIM 154705)
- MASS - myopia, mitral valve prolapse, mild aortic dilatation, skin and skeletal (at least 2, preferable 3 criteria)
- Familial mitral valve prolapse syndrome
- Stickler syndrome (MIM 108300) - myopia, vitreoretinal degeneration, retinal detachment, deafness arthropathy, spondyloepiphysyal dysplasia, joint hypermobility, midface hypoplasia, micrognathia, U-shaped cleft palate, mitral valve prolapse
- Shprintzen-Goldberg syndrome (MIM 182212) - Marfan type skeletal involvement AND craniosynostosis AND development delay

Notes
2. Wrist sign - thumb overlaps the distal phalanx of the fifth digit when grasping the contralateral wrist.
3. Thumb sign - entire nail of the thumb projects beyond the ulnar border of the hand when the hand is clenched without assistance.